ANALYTE:
NR5A1

NAME:	nuclear receptor subfamily 5 group A member 1
SYMBOL:	NR5A1
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	AD4BP ELP FTZ1 SF-1 SF1 hSF-1 steroidogenic factor 1
XREF(S):	Orphanet Ensembl Genatlas HGNC OMIM Reactome SwissProt
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RELATED CONTENT

Related Genetic Tests

- Congenital malformation (gene panel - 1721 genes)
- Congenital malformation gene panel
- Disorders of sex development - Primary Ovarian insufficiency - Hypogonadotropic Hypogonadism (gene panel)
- Intellectual disability (gene panel)
- Intellectual disability (virtual gene panel)
- Neurodevelopmental disorders (1300 genes)
- Neurodevelopmental disorders gene panel
- Premature Ovarian Failure/Primary Ovarian Insufficiency (POF/POI) (32 genes)
- Skin disorders (gene panel)

Related Diseases

- 46,XX gonadal dysgenesis
- 46,XX ovotesticular difference of sex development
- 46,XX testicular difference of sex development
- 46,XY complete gonadal dysgenesis
- 46,XY partial gonadal dysgenesis
- Male infertility with azoospermia or oligozoospermia due to single gene mutation
- NON RARE IN EUROPE: Primary ovarian failure

Related Gene Panels

- Congenital malformation (1721 genes) - ULB
- Congenital malformation gene panel - VUB

- Disorders of Sex Development - Primary Ovarian Insufficiency - Hypogonadotropic Hypogonadism - UGent
- Intellectual disability (gene panel)
- Intellectual disability/Epilepsy (1091 genes) - ULG
- Neurodevelopmental disorders (1300 genes) - ULB
- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Premature Ovarian Failure/Insufficiency (32 genes) - VUB
- Skin disorders - UGent
